

Title (en)
ATAXIA-TELANGIECTASIA GENE AND ITS GENOMIC ORGANIZATION

Title (de)
ATAXIA-TELANGIECTASIA GEN UND DESSEN GENOMISCHE ORGANISATION

Title (fr)
GENE DE L'ATAXIA-TELANGIECTASIE ET SON ORGANISATION GENOMIQUE

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Abstract (en)
[origin: US5858661A] A purified and isolated gene, designated ATM, mutations of which cause ataxia-telangiectasia and its genomic organization is disclosed. Methods and a kit for the detection of carriers of mutations of the ATM gene are provided by analysis of nucleic acids isolated from patients including in situ hybridization, Northern blotting and reverse transcriptase-polymerase chain reaction, Southern blotting, single strand conformational polymorphism, restriction endonuclease fingerprinting (REF), PCR amplification and DNA-chip analysis.

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